Name_____ Class_____ Date_____

CHROMOSOMES AND GENES

Although Mendel worked out many of the basic facts of heredity, he did not know about chromosomes or genes. Mendel worked in the 1860s, but it was not until the early 1900s that biologists rediscovered his work and made the connection between chromosomes and the units of heredity hypothesized by Mendel.

CHROMOSOME THEORY

In 1903, the American geneticist Walter Sutton presented his chromosome theory of heredity. He arrived at this theory after reviewing Mendel's work and drawing comparisons between Mendel's hypothetical units of heredity and the behavior of chromosomes during meiosis and fertilization. According to Sutton's theory, the units of heredity—the *genes*—are located on the chromosomes. In diploid cells, the chromosomes and genes are present in pairs. Pairs of similar chromosomes are called *homologous chromosomes*. Homologous chromosomes are made up of genes that influence the same traits and are located at identical positions on both chromosomes. Pairs of similar genes are called *alleles*.

During meiosis, chromosomes segregate, with one chromosome of each homologous pair passing to each gamete. Since pairs of alleles are located on pairs of homologous chromosomes, segregation of chromosomes results in the segregation of genes, with each daughter cell receiving one of each pair of alleles. The segregation of genes during meiosis and the recombination of genes during fertilization are causes of variation among offspring in sexual reproduction.

Questions

1. The units of heredity, which are called _____, are located on the _____.

2. What happens to each set of alleles in the course of meiotic cell division?

LINKAGE

According to Mendel's law of independent assortment, the hereditary factors are segregated independently of one another. However, in the course of experiments with both plants and fruit flies, investigators in the early 1900s found that certain traits appeared to be passed on together rather than independently. Further work showed that chromosomes are segregated independently, but that genes that are present on the same chromosome are generally passed on together. Such genes are said to show *linkage*.

Questions

1. Under what circumstances are genes inherited together?

2. How does the concept of linkage modify Mendel's law of independent assortment?

CROSSING OVER

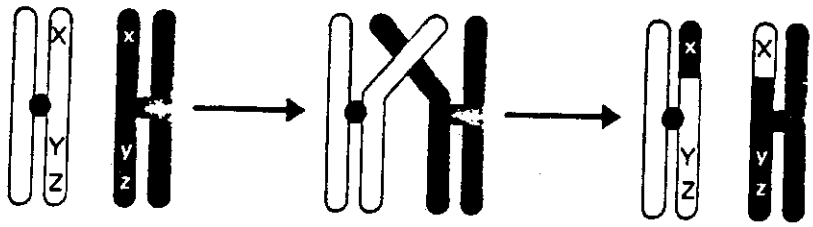
During meiosis, homologous pairs of chromosomes undergo synapsis (form tetrads). Sometimes there is an exchange of segments between the chromatids of a tetrad. In such an exchange of parts some linked genes become separated. This exchange of parts between homologous chromosomes is called *crossing over*. The greater the distance between two genes on a chromosome, the more likely they are to be separated by crossing over.

Questions

1. What is crossing over? At what stage in meiosis does it occur?

2. If two genes are at opposite ends of a chromatid, are their chances of being separated by crossing over greater or less than if they were close together?

3. Answer the following questions based on the diagrams below.



a. What is the relationship between genes X and x?

b. Which genes are exchanged by crossing over?

c. What new linkage groups are formed by the crossing over?

SEX CHROMOSOMES

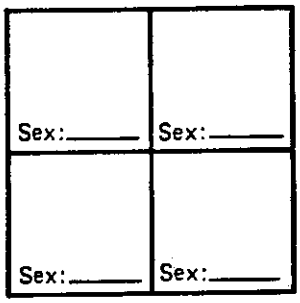
sex determination

Sex in humans and various other organisms (both plant and animal) is determined by a specific pair of chromosomes called the *sex chromosomes*. All chromosome pairs other than the sex chromosomes are called *autosomes*. In humans, the sex chromosomes are designated as X and Y. Females have two X chromosomes, while males have one X and one Y chromosome. In gamete formation, the eggs of the female always contain an X chromosome. However, two types of sperm are produced—one con-

taining an X chromosome and the other containing a Y chromosome. Thus, at fertilization, if the egg is fertilized by an X-containing sperm, the offspring will be female (XX). If the egg is fertilized by a Y-containing sperm, the offspring will be male (XY). In humans it is the sperm of the male that determines the sex of the offspring. In birds, on the other hand, it is the female who produces two types of gametes and thus determines sex.

Questions

1. In humans, there are _____ pair(s) of autosomes and _____ pair(s) of sex chromosomes.
2. The sex chromosomes in the human female are represented by _____ and in the human male by _____.
3. Use the Punnett square below to show the segregation of human sex chromosomes in the formation of gametes. Fill in the square showing all possible crosses. Give the phenotype (sex) of the offspring.



4. A mating of a pair of fruit flies produces 300 offspring. Approximately how many males and how many females will there be in this group?

sex-linked traits

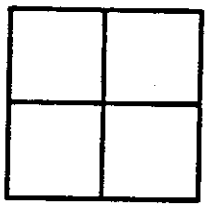
Traits determined by genes located on the X chromosome are said to be *sex-linked*. A number of undesirable sex-linked traits have been discovered in humans. One of the most common is color blindness. Hemophilia, a condition in which the blood does not clot properly, is another. These sex-linked traits are much more common in males than in females. This is because in humans and various other species the X chromosomes carry an assortment of genes, while the Y chromosomes have very few genes. Thus, when a gene is carried on the X chromosome of a male, there is no allele (corresponding gene) on the Y chromosome. Therefore, a gene on the X chromosome of the male will always be expressed, whether it is dominant or recessive. The genes for many of the undesirable sex-linked traits are recessive and relatively rare. When such a gene is present in a female, there is usually a dominant, normal allele on the other X chromosome that produces a normal phenotype. Although such females appear normal, they are *carriers* of the trait, which may be passed on to their offspring.

Questions

1. What is a sex-linked trait?
2. Two sex-linked traits in humans are _____ and _____.
3. In the diagrams below X represents the normal gene and X^- the defective gene. Label each diagram as male or female and as normal, afflicted (showing the trait of the defective gene), or carrier.

_____ _____ _____ _____ _____

4. Using the Punnett square below, show the possible offspring produced by a normal father (XY) and a mother who is a carrier for color blindness (XX^-).

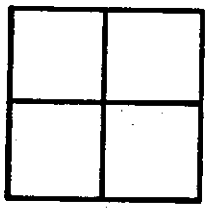


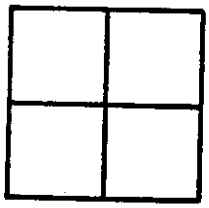
a. What percentage of their daughters will be color-blind? _____

b. What percentage of their daughters will be carriers of the gene for color blindness? _____

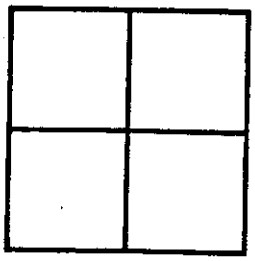
c. What percentage of their sons will be color-blind? _____

5. Using Punnett squares to work out your answers, describe the pattern of color blindness among the offspring of a color-blind mother and a normal father. Do the same for a color-blind mother and color-blind father.





6. Like color blindness in humans, white eyes in fruit flies is a sex-linked characteristic. Using a Punnett square, show the results of a cross between a white-eyed female (X^-X^-) and a red-eyed male (XY). In each square, label each of the offspring as red-eyed or white-eyed and, where appropriate, as a carrier.



Name_____ Class_____ Date_____

MULTIPLE ALLELES

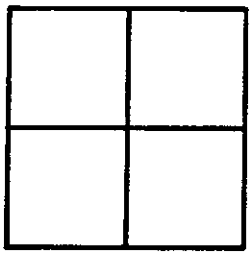
All traits discussed so far have been controlled by a single pair of alleles. However, there are some traits, such as blood type in humans, that involve more than two genes. Human blood type involves three genes—A, B, and O. Genes A and B are dominant, while O is recessive. The four human blood types are A, B, AB, and O. Genotypes of multiple alleles are shown using the capital letter I for dominant and lower case i for recessive. Thus, for example, type A blood is genotype I^A.

Questions

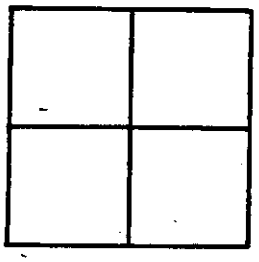
1. What are multiple alleles?

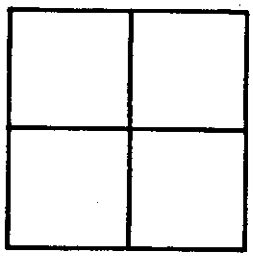
2. Two traits in humans that are determined by multiple alleles are _____ and _____.

3. A man with type AB blood marries a woman with type O blood. Using the Punnett square below, show all possible genotypes of their offspring.



4. Could a mother with type B blood and a father with type O blood produce offspring with type AB blood? Use the Punnett squares below to check your answer.





MUTATIONS

Mutations are stable changes in the hereditary material. *Somatic mutations* are genetic changes within the body cells. This type of mutation can be passed on only to other body cells by mitotic cell division. *Germinal mutations*, which are mutations in the sex, or *germ*, cells, can be passed on to succeeding generations.

Any trait can undergo a mutation at any time, but the rate of mutation is usually very low. Certain types of radiation and various chemicals are known to cause mutations. Mutations are generally recessive. Thus, they are usually masked by dominant genes unless they are sex-linked. Muta-

tions are generally disadvantageous. They tend to cause disruptions in the biochemical machinery of the cell.

Chromosome mutations involve permanent changes in chromosome structure. These changes usually occur during meiosis. In *crossing over* there is an exchange of chromosome segments between homologous chromosomes. *Inversions* involve the rotation of a piece of chromosome so that the order of genes in that segment is reversed. *Duplication* of chromosome segments occurs when a segment broken off one chromosome becomes attached to the homologous chromosome, resulting in a duplication of some genes. In *deletion*, a segment breaks off a chromosome, resulting in the loss of some genes. In *translocation*, a segment from one chromosome is transferred to a nonhomologous chromosome.

Gene mutations, which involve changes in DNA, are discussed in Unit 19.

Questions

1. What is a mutation?

2. Changes in chromosomes of body cells are called _____ mutations.
3. Changes in the chromosomes of gametes are called _____ mutations.
4. Name four types of chromosome mutations.

polyploidy

During the normal process of meiosis, the sets of doubled chromosomes separate, with one of each chromosome going to each daughter cell. Occasionally, the doubled chromosomes do not separate, resulting in a cell with more than the diploid number of chromosomes. The condition in which the cells of an organism have more than twice the haploid number of chromosomes is called *polyploidy*. Plants carrying the tetraploid ($4n$) chromosome number are more vigorous and larger than the diploids. Many fruits sold commercially are polyploids. The chemicals colchicine and camphor are used to induce polyploidy in some plants. Polyploidy is rarely found in animals because the condition is usually lethal.

Question

The condition in which the cells of an organism have more than twice the haploid number of chromosomes is called _____.

nondisjunction

When a pair of homologous chromosomes does not separate during meiotic cell division, the abnormality is called *nondisjunction*. If nondisjunction occurs in only a single chromosome pair, the result is a gamete with one extra chromosome ($n + 1$). If this gamete is involved in fertilization, the resulting zygote will have a chromosome number of ($2n + 1$). In humans, nondisjunction of chromosome pair 21 results in *mongolism*, or *Down's syndrome*.

Question

The failure of homologous chromosomes to separate during meiosis is called _____.